

## Ⅱ 研究成果の刊行に関する一覧表



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発表者名	論文タイトル名	発表雑誌	巻号	ページ	出版年
Moriya K, Suzuki T, Uchida N, Nakano T, Katayama S, Irie M, Rikiishi T, Niizuma H, Okada S, Imai K, Sasahara Y, Kure S.	Ruxolitinib treatment of a patient with steroid-dependent severe autoimmunity due to STAT1 gain-of-function mutation.	Int J Hematol.	in press		2020
Moriya K, Kadowaki S, Nakano T, Kutukculer N, Aksu G, Sasahara Y, Kure S, Ohnishi H, Jean-Laurent Casanova JL, Puel A, Fukao T.	The IL1RN mutation creating the most-upstream premature stop codon is hypomorphic because of a reinitiation of translation.	J Clin Immunol.	in press		2020
Sato S, Ohnishi T, Uejima Y, Furuichi M, Fujinaga S, Imai K, Nakamura K, Kawano Y, Suganuma E.	Induction therapy with rituximab for lupus nephritis due to prolidase deficiency.	Rheumatology (Oxford).	in press		2020
Umeda K, Imai K, Yanagimachi M, Yabe H, Kobayashi M, Takahashi Y, Kajiwara M, Yoshida N, Cho Y, Inoue M, Hashii Y, Atsuta Y, Morio T. Inherited Disease Working Group of the Japan Society for Hematopoietic Cell Transplantation.	Impact of graft-versus-host disease on the clinical outcome of allogeneic hematopoietic stem cell transplantation for non-malignant diseases.	Int J Hematol.	in press		2020
Dezfouli M, Bergström S, Nonoyama S, Hammarström L, et al.	Newborn Screening for Presymptomatic Diagnosis of Complement and Phagocyte Deficiencies.	Front Immunol.	11	455	2020
Nochi T, Suzuki S, Ito S, Morita S, Furukawa M, Fuchimoto D, Sasahara Y, Usami K, Niimi K, Kitago M, Matsuda S, Matsuo A, Suya, a Y, Sakai Y, Wu G, Bazer FW, Watanabe K, Onishi A, Aso H.	Elucidation of the effects of a current X-SCID therapy on intestinal lymphoid organogenesis using an in vivo animal model.	Cell Mol Gastroenterol Hepatol.	10(1)	83-100	2020
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Murakami N, Sakai T, Arai E, Muramatsu H, Ichikawa D, Asai S, Shimoyama Y, Ishiguro N, Takahashi Y, Okuno Y, Nishida Y.	Targetable driver mutations in multicentric reticulohistiocytosis.	Haematologica.	105(2)	e61-e64	2020
Iwata Y, Satou K, Furuichi K, Yoneda I, Matsumura T, Yutani M, Fujinaga Y, Hase A, Morita H, Ohta T, Senda Y, Sakai-Takemori Y, Wada T, Wada T, et al.	Collagen adhesion gene is associated with bloodstream infections caused by methicillin-resistant Staphylococcus aureus.	Int J Infect Dis.	91	22-31	2020
Iwata Y, Sakai N, Yoneda I, Satou K, Furuichi K, Senda Y, Sakai-Takemori Y, Wada T, Wada T, et al.	The increased frequency of methicillin-resistant Staphylococcus aureus with low MIC of beta-lactam antibiotics isolated from hospitalized patients.	J Infect Chemother.	26(6)	604-610	2020
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Yokoyama K, Horiuchi T, Hashimura C, Yoshida A.	A novel C1 inhibitor gene mutation in a family with hereditary angioedema: Use of genetic analysis to facilitate early diagnosis.	Allergol Int.	69(1)	148-149	2020
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Yoshikawa T, Ihira M, Higashimoto Y, Hattori F, Miura H, Sugata K, Komoto S, Taniguchi K, Iguchi A, <u>Yamada M</u> , <u>Ariga T</u> .	Persistent systemic rotavirus vaccine infection in a child with X-linked severe combined immunodeficiency.	J Med Virol.	91	1008-1013	2019
Kobayashi I, Okura Y, Ueki M, Tozawa Y, Takezaki S, <u>Yamada M</u> and <u>Ariga T</u> .	Evaluation of systemic activity of pediatric primary Sjögren's syndrome by EULAR Sjögren's syndrome disease activity index (ESSDAI).	Mod Rheumatol.	29	130-133	2019
Ueki M, Kobayashi I, Takezaki S, Tozawa Y, Okura Y, <u>Yamada M</u> , Kuwana M and <u>Ariga T</u> .	Myositis-specific autoantibodies in Japanese patients with juvenile idiopathic inflammatory myopathies.	Mod Rheumatol.	29	351-356	2019
Okamura K, Uchida T, Hayashi M, Yaguchi Y, Hemmi A, Murata I, Ichikawa K, Koyama S, Onoda T, <u>Sasahara Y</u> , Suzuki T.	Neutrophilic dermatosis associated with NFKB2 mutation.	Clin Exp Dermatol.	44(3)	350-352	2019
Umeda K, Yabe H, Kato K, <u>Imai K</u> , Kobayashi M, Takahashi Y, Hama A, Inoue M, <u>Sasahara Y</u> , Kato K, Adachi S, Koga Y, Hara J, Hashii Y, Atsuta Y, <u>Morio T</u> ; on behalf of the Inherited Disease Working Group of the Japan Society for Hematopoietic Cell Transplantation.	Impact of low-dose irradiation and in vivo T-cell depletion on hematopoietic stem cell transplantation for non-malignant diseases using fludarabine-based reduced-intensity conditioning.	Bone Marrow Transplant,	54(8)	1227-1236	2019
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Tsutsumi N, Yokota A, Kimura T, Kato Z, Fukao T, Shirakawa M, <u>Ohnishi H</u> , Tochio H.	An innate interaction between IL-18 and the propeptide that inactivates its precursor form.	Sci Rep.	9(1)	6160	2019
Nozawa A, Ozeki M, Matsuoka M, Nakama M, Yasue S, Endo S, Kawamoto N, <u>Ohnishi H</u> , Fukao T.	Perampanel Inhibits Neuroblastoma Cell Proliferation Through Down-regulation of AKT and ERK Pathways.	Anticancer Res.	39(7)	3595-3599	2019
Hori T, <u>Ohnishi H</u> , Kadowaki T, Kawamoto N, Matsumoto H, <u>Ohara O</u> , Fukao T.	Autosomal dominant Hashimoto's thyroiditis with a mutation in TNFAIP3.	Clin Pediatr Endocrinol.	28(3)	91-96	2019
Ibusuki A, Nishikawa T, Hiraki T, Okano T, <u>Imai K</u> , <u>Kanegane H</u> , <u>Ohnishi H</u> , Kato Z, Fujii K, Tanimoto A, Kawano Y, Kanekura T.	Prominent dermal Langerhans cells in an Omenn syndrome patient with a novel mutation in the IL2RG gene.	J Dermatol.	46(11)	1019-1023	2019
Nozawa A, Ozeki M, Yasue S, Endo S, Kawamoto N, <u>Ohnishi H</u> , Fumino S, Furukawa T, Tajiri T, Maekawa T, Fujino A, Souzaki R, Fukao T.	Immunologic Effects of Sirolimus in Patients With Vascular Anomalies.	J Pediatr Hematol Oncol.	in press		2019
Ogawa B, Aoki M, <u>Ohnishi H</u> , Ohashi T, Hayashi H, Kuze B, Ito Y.	The Long-Term Efficacy of Cochlear Implantation for Hearing Loss in Muckel-Wells Syndrome.	J Int Adv Otol.	15(3)	454-458	2019
Mori M, Hira A, Yoshida K, <u>Muramatsu H</u> , Okuno Y, Shiraishi Y, Anmae M, Yasuda J, Tadaka S, Kinoshita K, Osumi T, Noguchi Y, Adachi S, Kobayashi R, Kawabata H, <u>Imai K</u> , <u>Morio T</u> , Tamura K, Takaori-Kondo A, Yamamoto M, Miyano S, Kojima S, Ito E, Ogawa S, Matsuo K, Yabe H, Yabe M, Takata M.	Pathogenic mutations identified by a multimodality approach in 117 Japanese Fanconi anemia patients.	Haematologica.	104 (10)	1962-1973	2019

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Muraoka M, Akagi T, Ueda A, <u>Wada T</u> , Koefler H.P, Yokota T, Yachie A.	C/EBPε ΔRS derived from a neutrophil-specific granule deficiency patient interacts with HDAC1 and its dysfunction is restored by trichostatin A.	Biochem Biophys Res Commun.	516	293-299	2019
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Hosaka S, Kobayashi C, Saito H, Imai-Saito A, Suzuki R, Iwabuchi A, Kato Y, Jimbo T, Watanabe N, <u>Onodera M</u> , Imadome KI, Masumoto K, Nanmoku T, Fukushima T, Kosaki K, Sumazaki R, <u>Takada H</u> .	Establishment of immunity against Epstein-Barr virus infection in a patient with CHARGE/complete DiGeorge syndrome after peripheral blood lymphocyte transfusion.	Pediatr Transplant.	23	e13424	2019
Tomono T, Hirai Y, Chono H, Mineno J, Ishii A, <u>Onodera M</u> , Tamaoka A, Okada T.	Infectivity Assessment of Recombinant Adeno-Associated Virus and Wild-Type Adeno-Associated Virus Exposed to Various Diluents and Environmental Conditions.	Hum Gene Ther Methods.	30	137-143	2019
Kikuchi T, Nakae J, Kawano Y, Watanabe N, <u>Onodera M</u> , Itoh H.	Foxo in T Cells Regulates Thermogenic Program through Ccr4/Ccl22.	iScience	22	81-96	2019
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Maeshima K, <u>Okada S</u> , Shibata H. Dr. Maeshima, et al.	reply.  Comment on Maeshima K, Ishii K, Shibata H. An Adult Fatal Case with a STAT1 Gain-of-function Mutation Associated with Multiple Autoimmune Diseases. J Rheumatol. 2019 Mar;46(3):325-327.  Goulielmos GN, Zervou MI, Eliopoulos E. Functional Significance of the C324R Mutation Examined Using a Structural Biological Approach. J Rheumatol. 2019 Jun;46(6):654-655.	J Rheumatol.	46(6)	655-656	2019
Niihori T, Nagai K, Fujita A, Ohashi H, Okamoto N, <u>Okada S</u> , Harada A, Kihara H, Arbogast T, Funayama R, Shirota M, Nakayama K, Abe T, Inoue SI, Tsai IC, Matsumoto N, Davis EE, Katsanis N, Aoki Y.	Germline-Activating RRAS2 Mutations Cause Noonan Syndrome.	Am J Hum Genet.	104(6)	1233-1240	2019

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Kumaki E, Tanaka K, <u>Imai K</u> , Aoki-Nogami Y, Ishiguro A, <u>Okada S</u> , <u>Kanegane H</u> , Ishikawa F, <u>Morio T</u> .	Atypical SIFD with novel TRNT1 mutations: a case study on the pathogenesis of B-cell deficiency.	Int J Hematol.	109(4)	382-389	2019
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Otani T, Hoshika S, <u>Horiuchi T</u> , Hashimura C, Sugihara S.	Hereditary angioedema: Repeated attacks in a 10-year-old boy.	Pediatr Int.	61(1)	101-103	2019
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Shigemura T, Matsuda K, Kurata T, <u>Nakahata T</u> , Koike K. et al.	Essential role of PTPN11 mutation in enhanced haematopoietic differentiation potential of induced pluripotent stem cells of juvenile myelomonocytic leukaemia.	Br J Haematol.	187(2)	163-173	2019
Saeki S, Enokizono T, Imagawa K, Fukushima H, Kajikawa D, Sakai A, Tanaka M, Ohto T, Suzuki H, Uehara T, Takenouchi T, Kenjiro K, & <u>Takada H</u> .	A case of autism spectrum disorder with cleft lip and palate carrying a mutation in exon 8 of AUTS2.	Clinical case reports,	7(11)	2059–2063	2019
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Tozawa Y, Abdrabou SSMA, Nogawa-Chida N, Nishiuchi R, Ishida T, Suzuki Y, Sano H, Kobayashi R, Kishimoto K, Ohara O, <u>Imai K</u> , <u>Naruto T</u> , <u>Kobayashi K</u> , <u>Ariga T</u> , <u>Yamada M</u> .	A deep intronic mutation of c.1166-285 T>G in SLC46A1 is shared by four unrelated Japanese patients with hereditary folate malabsorption (HFM).	Clin Immunol.	208	108256	2019
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